

15. A method of identifying a presence of Down Syndrome in a fetus, comprising:
obtaining an amniotic fluid specimen by placing a syringe having a needle into a uterus
and withdrawing the amniotic fluid specimen via the needle,
identifying a quantity for each metabolite that is present in the amniotic fluid specimen
using a gas chromatograph/mass spectrometer,
compiling a patient profile, wherein the patient profile lists each metabolite and the
quantity for each respective metabolite,
comparing the patient profile with a control profile representative of normal levels of
each metabolite, wherein the control profile lists a quantity for each respective metabolite of the
patient profile that is present in amniotic fluid of persons with Down Syndrome, by comparing
the quantity of each metabolite of the patient profile with the quantity for that respective
metabolite of the control profile, and
identifying the presence of Down Syndrome in the fetus when a quantity of a subset of
metabolites of the patient profile has a different quantity than each respective metabolite of the
control profile.

21. A method of identifying a presence of Down Syndrome in a fetus, comprising:
obtaining an amniotic fluid specimen by placing a needle into a uterus and withdrawing
the amniotic fluid specimen via the needle,
identifying a quantity for each metabolite that is present in the amniotic fluid specimen
by analyzing the amniotic fluid specimen using a gas chromatograph/mass spectrometer,
compiling a patient profile, wherein the patient profile lists each metabolite and the
quantity for each respective metabolite present in the amniotic fluid specimen,

obtaining a control profile, wherein the control profile lists a quantity for each metabolite present in the amniotic fluid specimen for a population of patients without Down Syndrome.

identifying a plurality of abnormal quantities of metabolites of the patient profile by comparing the quantity of each metabolite of the patient profile with the quantity for that respective metabolite of the control profile, and

identifying the presence of Down Syndrome in the fetus when the plurality of abnormal quantities of metabolites of the patient profile corresponds to abnormal quantities of those metabolites in amniotic fluid of a patient known to have Down Syndrome.